Family Health History Multi-Stakeholder Workgroup

Data Requirements Summary

Presented to:

The Personalized Health Care Workgroup of the American Health Information Community

December 2007

Family Health History Multi-Stakeholder Workgroup Co-chairs:

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1.0 Introduction

On July 31, 2007, the Personalized Health Care (PHC) Workgroup (http://www.hhs.gov/healthit/ahic/healthcare/) submitted a set of recommendations to the America Health Information Community (AHIC). These recommendations, subsequently adopted by AHIC, were aimed at enhancing the integration of interoperable family health history information into Electronic Health Records (EHRs). Considering that there is not a universally accepted minimum set of family health history data to be collected in primary care, one of the recommendations was to develop a core minimum data set and common data definitions to properly collect family health history information.

Recommendation 3.0:

> A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to develop a core minimum data set and common data definition available for primary care collection of family health history information.(Is this a recommendation from the report? It needs more to establish its context)

A workgroup was formed in accordance with the recommendation. This document reports the result of the workgroup activity. The core data set was established through a consensus- building process involving a wide variety of stakeholders. The effort drew on expertise from both the public and private sector related to family health history in primary care, electronic health records, and health care delivery.

1.1 Purpose

The purpose of this document is to deliver the recommended core data set requirements for collecting family health history across multiple clinical agencies. Workgroup recommendations for both required data and optional data are included.

1.2 Scope

This document describes the project background, methodology, findings, and next steps. Additionally, it provides the requirements table including associated clarifications.

2.0 Background

Personalized Health Care is one of ten priories identified by Secretary Michael O. Leavitt, Department of Health and Human Services (HHS). The Secretary's vision for PHC is that "Personalized health care is information-based health care. It is health care that works better for each patient, based partly on scientific information that is new, and partly on technology to make complex information useful. Whether it involves new biomedical knowledge, data networks for developing that knowledge, or computer supports to manage that knowledge, personalized health care is about a transformed role for information in health care."

On September 13, 2005, Secretary Leavitt announced the membership for the AHIC. The AHIC was formed to help advance efforts to reach President Bush's call for most Americans to have electronic health records within ten years. The AHIC is a federally-chartered advisory committee and provides input and recommendations to HHS on how to make health records digital and interoperable, and assure that the privacy and security of those records are protected, in a smooth, market-led way.

The AHIC, chaired by Secretary Leavitt and Dr. Robert Kolodner, Director, Office of the National Coordinator for Health Information Technology, established seven workgroups that involve over 100 experts. Workgroups develop recommendations to the AHIC and subsequently to the Secretary for action. The Personalized Health Care Workgroup is one of these seven workgroups.

The PHC Workgroup's vision of PHC is a forward-looking, consumer-centric system in which clinicians customize diagnostic, treatment, and management plans based on a variety of factors, including culture, personal behaviors, preferences, family health history, and their unique genetic/genomic makeup. This vision is based on the confluence of two powerful forces, the development of Health Information Technology (HIT), and the rapid advances in our basic understanding of the relationships between health, disease, and genetics. The PHC Workgroup's specific charge is to:

> Make recommendations to the AHIC to consider means to establish standards for reporting and incorporation of common medical genetic/genomic tests and family health history data into electronic health records, and provide incentives for adoption across the country including federal government agencies.

One of the four priority areas identified by the PHC Workgroup is family health history. Health care professionals and the general public have widely accepted the importance of family health history for assessing risk for a number of common diseases, including cancer, heart disease, and diabetes. Despite this wide acceptance, there is a paucity of Personal Health Record (PHR) or EHR systems capable of capturing family health history data in a structured, standardized and interoperable format that can be integrated seamlessly with electronic clinical decision support (CDS) tools.

As our scientific understanding of the molecular and genetic/genomic basis for health and disease improves, the importance of family health history as a predictive tool has increased. This fact has been highlighted throughout HHS by the Surgeon General's online web portal for collecting family health history information, 'My Family Health Portrait', developed in conjunction with the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH) and the Centers for Disease Control and Prevention (CDC). This tool is similar to a PHR in that its use and data entry are all patient-driven. The Family Health History priority area for the PHC Workgroup includes activities of immediate concern related to use case development by the Office of the National Coordinator for Health IT (ONC). Upon completion of the use cases by ONC, after several rounds of public comment, the use cases are then passed on the Health Information Technology Standards Panel (HITSP). The mission of the HITSP is to serve as a cooperative partnership between the public and private sectors for the purpose of achieving a widely accepted and useful set of standards specifically to enable and support widespread interoperability among health care software applications, as they will interact in a local, regional, and national health information network for the United States. The use case should represent the continuum of information collection, from consumer entry of family health history in the PHR to clinician entry of family health history in the EHR. The long term goal is the interoperability between the PHR and EHR. In order to support the use case development process there is a clear need to develop a core minimum set of data and common data definitions for the collection of family health history information.

3.0 Methodology

The Family Health History co-chairs assembled a multi-stakeholder workgroup comprised of over 40 members representing approximately 18 different organizations. The workgroup was first convened shortly after the PHC Workgroup Recommendation 3.0 was approved on July 31, 2007.

Initially, a straw-man document was circulated to the workgroup. This straw-man document was based on a draft of proposed standards for family health history information developed for the Continuity of Care Record (CCR). The straw-man document generated a discussion focused on what the stakeholders felt to be the core set of family health history information in the primary care health delivery environment. Stakeholders were then asked to supply comments on the CCR document. In addition, stakeholders were asked for relevant materials their organizations used internally to define the core data set for family health history. The comments and the supporting documents supplied by the stakeholders were then used to assemble a draft document that defined the core variables and functionalities related to the representation of family health history information in the EHR/PHR. Stakeholders then engaged in a process whereby they designated these elements and functionalities as required or optional in the EHR/PHR environment. They were asked to consider several factors when designating a concept or function as required or optional. These were:

a) the perspective was that of the use of family health history information by primary care providers or patient in the EHR/PHR,
b) the concepts or functions that the EHR/PHR needed to be able to capture or perform, and that the health care provider (or patient in the case of a PHR) may or may not use all of these concepts or functions in any given encounter, and
c) the listing should include concepts and functions that would be relevant to EHR/PHR users today and 5-10 years into the future.

Stakeholder responses were reviewed by the task force chairs, summarized, and used as a basis for three facilitated phone discussions among the stakeholders to achieve consensus on the family health history concepts/functionalities that should be part of every EHR/PHR.

3.1 Reference Documents

The following references were utilized for domain research and project background:

3.2 Multi-Stakeholder Workgroup

Eighteen different organizations are represented in the Family Health History Multi-Stakeholder Workgroup. This group includes members from the private sector and federal agencies who have been active in the PHC Workgroup or subgroups as well as individuals with knowledge and

expertise in the area of family health history, EHRs, and health care delivery. In particular, an effort was mate to include representatives from private sector organizations that would be affected by the inclusion of a family helath history core data set in the electronic health record. Since August 2007 the workgroup has had three conference calls and numerous email exchanges to define the minimum core data set.

The following tables identify the members of the multi-stakeholder workgroup:

Table 1 Federal Partners

Federal Partners		
Name	**Position**	**Agency**
Mary Beth Bigley, DrPH, MSN, ANP	Senior Health Fellow	Office of the Surgeon General
Kristin Brinner, Ph.D.	2006-2008 AAAS Fellow	Personalized Health Care Initiative
Terry Cullen, M.D., MS	Chief Information Officer/Director of Office of Information Technology	Indian Health Service
Nhan V Do, M.D., MS, FACP LTC, MC	Chief, Medical Informatics	Tricare Management Activity Department of Defense
Greg Feero, M.D., Ph.D.	Chief, Genomic Healthcare Branch	National Human Genome Research Institute
Linda Fischetti, RN, MS	Chief Health Informatics Officer	Veterans Health Administration
Emory Fry, M.D.		Department of Defense
Alan Guttmacher, M.D.	Deputy Director	National Human Genome Research Institute
Theresa Hancock, PAHM	Acting Director, Veterans/ Consumer Health Informatics Office	Veterans Health Administration
Betsy Humphreys, M.D.	Deputy Director	National Library of Medicine
Nelson Hsing, Sc.D., MHS	Management Analyst Health*e*People	Veterans Health Administration
Katie Kolor, PhD, Ms, CGC	Policy Officer	National Office of Public Health Genomics, CDC

Federal Partners		
Name	**Position**	**Agency**
Joel Kupersmith, M.D.	Chief Research and Development Officer, Office of Research and Development	Department of Veterans Affairs
Penny Kyler, MA, OTR, FAOTA	Genetic Services Branch	Maternal and Child Health Bureau, HRSA
Michele Lloyd-Puryear MD, Ph.D.	Chief, Genetic Services Branch	Maternal and Child Health Bureau, HRSA
Marie Mann, M.D., M.P.H.	Genetic Services Branch	Maternal and Child Health Bureau, HRSA
Clem McDonald, M.D.,	Director of Lister Hill Center	National Library of Medicine, National Institutes of Health
Scott McLean, MD COL, MC, United States Army	Chief, Medical Genetics	San Antonio Military Medical Centers - BAMC/WHMC Clinical Genetics Consultant to the Army Surgeon General
Paul Nickel, MD	National Director Medical Informatics, PCS Office of Patient Care Services	Veterans Health Administration
Timothy O'Leary, MD, PhD	Acting Director of CSRD	Veterans Health Administration
David Parker, BS, BSN, MHS	Business Systems Analyst	Indian Health Service/DNC
Rodolfo Valdez, PhD, MSc	Epidemiologist	National Office of Public Health Genomics CDC
Daniel Wattendorf, MD	Director, Air Force Medical Genetics Center	United States Air Force

Table 2 Non-federal Partners

Non-federal Partners		
Name	**Position**	**Agency**
Cephus Daniel V. Allin, MD	Physician Consultant	NextGen Healthcare Information Systems
Mike Brammer	Chief Executive Officer	Progeny Software, LLC
Andrew Brown, Ph.D. FRSA	Director of Business Development	Progeny Software, LLC
Sarah Corley, MD, FACP	Chief Medical Officer	NextGen Healthcare Information Systems, Inc
Michael Crouch, M.D., MPH	Baylor Family Medicine Center, Department of Family and Community Medicine	Society of Teachers of Family Medicine
Ardis Davis, MSW	Executive Director	Association of Departments of Family Medicine
Ronald Dionne, M. D.	Product Manager	Allscripts
Debra Lochner Doyle, MS, CGC	State Genetics Coordinator	Washington State Department of Health
Matthew Ferrante	President	Primetime Medical Software
Tabitha Harrison, MPH	Genetics Services Specialist	Washington State Department of Health
Don Heim, PMP	Senior Research Scientist	Battelle Arlington Operations
Mark Hoffman, Ph.D., B.A.	Director - Translational Medicine	Cerner Corporation
Kevin Hughes, M.D., FAC	Surgical Director, Breast Screening, Co-Director,	Avon Comprehensive Breast Evaluation Center Massachusetts General Hospital
Dave Lareau	Chief Operating Officer	Medicomp Systems (Medcin)
Maki Moussavi, B.S. , MS	Genomics Strategist	Cerner Corporation
Maren T. Scheuner, MD, MPH, FACMG	Natural Scientist	RAND Corporation Adjunct Associate Professor, UCLA School of Public Health
Roger Sherwood	Executive Director	Society of Teachers of Family Medicine

Non-federal Partners		
Name	**Position**	**Agency**
Amnon Shabo, PhD	Research Staff Member	IBM Research Lab in Haifa
Mollie Ullman-Cullere MS, MSE	Senior Information Architect/Project Manager	Harvard Medical School - Partners Healthcare Center for Genetics and Genomics
Steven E. Waldren, M.D., M.S.	Director, Center for HIT	American Academy of Family Physicians
Marc Williams, MD, FAAP, FACMG	Director, Clinical Genetics Institute	Intermountain Healthcare, Salt Lake City, UT
Grant Wood	Senior IT Strategist	Intermountain Healthcare

Table 3 Continuity of Care Record Family History Workgroup

Continuity of Care Record Family History Workgroup **(contributors to the CCR document used in the draft of this report)**		
Name	**Position**	**Agency**
Louise Acheson, M.D., M.S.	Associate Professor of Family Medicine and Oncology and Assistant Professor of Reproductive Biology	Case Western Reserve University
Tom Agresta, M.D.	Associate Professor, Department of Family Medicine	University of Connecticut School of Medicine
Alexander Blount, Ed.D.	Professor of Clinical Family Medicine	University of Massachusetts Medical School
Greg Feero, M.D., Ph.D.	Chief, Genomic Healthcare Branch	National Human Genome Research Institute
Robert Gramling, M.D.	Assistant Professor, Family Medicine and Community Health	Brown University
Caryl Heaton, D.O.	Associate Professor, Department of Family Medicine	University of Medicine and Dentistry of New Jersey
Susan McDaniel, Ph.D.	Professor of Psychiatry and Family Medicine	University of Rochester

Nancy Stevens, M.D., M.P.H	Professor of Family Medicine	University of Washington

4.0 Summary of Findings

The workgroup recommends specific family health history data to be captured, stored and viewed in both EHR and PHR environments, and has additionally documented desired functionality for a family health history module. The workgroup recognized some disparities regarding the level of detail needed for the patient, or person of focus, as opposed to data pertaining to family members of the patient. These differences are annotated in the requirements table found in section 6 of this document.

Additionally, the workgroup intentionally omitted the finer details regarding the specific format for data collection. For example, many recommended data entry fields could potentially be programmed with canned or "pick list" responses as opposed to free text. The workgroup felt it is appropriate to allow each implementing agency to determine the format for data entry, recognizing that canned responses and data structures may differ among the various legacy systems.

Finally, the workgroup understands that much of the recommended core data set may already exist in legacy systems and EHRs. Implementing agencies are therefore encouraged to "pull" family health history data that already exists rather than duplicate entry and storage of data in a family health history module.

5.0 Future Directions

Family health history is a complex, multifaceted tool for assessing disease risk. Ultimately, it can be a tool for gaining an understanding of the interplay between inherited and social factors that are relevant to the care of patients. The value of a family health history tool in the EHR/PHR environment resides in enabling the user to collect, represent, and interpret structured data obtained from patients and other sources in a manner that properly supports clinical decisions. Further, the main goal will be achieved if this structured data can be made interoperable between entities, which ensure the availability of important medical information to all health care entities involved in the continuum of patient care with minimal duplication of effort.

This document will be submitted for consideration during the use case development process of AHIC as a potential benchmark for the family health history content of EHR/PHR systems seeking standards development and certification. At the same time, we hope that this document will provide guidance to entities that are developing or updating the family health history capabilities of their EHR/PHR systems.

Clearly much work is needed to translate this document, which largely deals with the conceptual content of family health history in the EHR/PHR, to an interoperable approach to capture, store, and link all the encompassed concepts that emerge as patients relate to their health care providers.

6.0 Requirements Table

The requirements table encompasses the recommended minimum core data set for family health history and includes basic desired functionality. The column headings in the table are defined as follows:

- **ID:** Unique requirement identifier used to reference a requirement.

- **Focus:** There are two focus areas of interest; the "Individual" and the "Family". In general, the term "Individual" refers to any person represented in the family health history obtained by the clinician or provided by the patient, including the individual who is the focus of the history. In most cases this person will be the actual or potential patient; also referred to as the index case (or proband). The group felt it was necessary to make a distinction between data pertaining to the patient and data pertaining to the family members for a few data elements. In general, the term "Family" refers to the biological relatives of the patient who is providing the family health history. The group recognized that "Family" actually encompasses more than simply the biological relationships between individuals. However, the group also felt that the primary focus of its work should be to develop the core data set that would be useful to automated clinical decision support for disease risk assessment in the primary care environment. Future efforts may be able to take a more integrated view of family health history as it relates to the social environment and health risks.

- **Requirement:** The requirement statement. This statement includes either the data collection requirement, high level functionality, or a combination of both.

- **Clarification:** A statement that clarifies the requirement and may include examples and/or additional detail.

- **Required:** This column indicates whether the workgroup recommends the requirement should be enforced as part of the core minimum data set for both an EHR and PHR. A "Yes" in this column does not necessarily mean data entry is forced in the applicable field, but that the data field should be available. Decisions regarding forced data entry should be left up to the implementing agencies. A "No" in this column indicates the workgroup recommends the requirement should be optional in an EHR environment, and in general, not applicable in a PHR environment.

- **Follow-Up:** This column contains information that will need to be considered or decisions made before implementation of the requirement can be accomplished.

Table 4 Family Health History Requirements

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
001	All	Data from existing legacy systems (EHR systems) shall be integrated and used to populate family history data fields.	Agencies shall not enter family history data that duplicates data already stored in a legacy system. Examples could include age, past medical history relevant to family history (thought to be critical for disease risk calculation), and self-described ethnicity data.	Various	Yes	
002	All	Free text fields shall be minimized for data entry of family history.	Canned or pick list data entry options should be utilized whenever possible to allow for maximum reporting functionality.	Various	Yes	Individual agencies will need to define canned data entry responses based on preference and industry standards.
003	Individual	Name shall be recorded.	For relatives, confidentiality may limit ability to represent this information.	First, Last	Yes	
004	Individual	Individual numerical identifier shall be recorded.	For patient and relatives. This number provides a means to distinguish individuals in order to delineate relationships. This would generally be assigned by the electronic system in which the data was collected, and would be used only for the purpose of structuring the family health history.	Identifier Number	Yes	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
005	Individual	Age shall be recorded.	For patient, full date of birth. For relatives, full date of birth is optimal, but due to identifiable nature, the ability to capture in this format may be limited. Minimum capture should be the year of birth. Less desirable would be the capture of the relative's age with a date stamp at time it is recorded so it can be updated accurately either automatically or manually.	Date of Birth/Year of Birth/Age	Yes	
006	Individual	Age of Death shall be recorded.	For relatives, age at death is sufficient.	Age at Death	Yes	
007	Individual	Cause of Death shall be recorded.	Include if known or note if unknown.	Cause of Death/Unknown Indicator	Yes	Agencies will need to decide if this should be a free text field, or canned responses.
008	Individual	Ethnicity/race shall be recorded.	Self defined for patient. Ethnicity/race/origins of grandparents should be collected and represented. This is a standard practice in construction of pedigrees by genetic professionals. However discussion reflected that multiple issues, including reliability of this information, make this approach problematic particularly from the perspective of a primary care provider.	Ethnicity/Race	Yes	
009	Individual	Biological Sex shall be recorded.		Sex (Male/Female)	Yes	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
010	Individual	Multiple-birth status shall be recorded.	Individual shall indicate if one of twins, triplets, etc, and whether identical or fraternal.	Multiple-birth status/Identical/ Fraternal	Yes	If a function is developed to distinguish twin status, it would be easy to apply this to any relative in the history if desired.
011	Individual	Biological parents shall be identified and recorded.	In the EHR/PHR environment, identification of biological parents for all relatives allows the construction of relationships which in aggregate form the backbone of the family health history and permit construction of a pedigree.	Biological parent's family health history data	Yes	
012	Individual	Consanguinity shall be identified and recorded.	Group consensus was that this concept should be represented for the parents of the patient.	Consanguinity	Yes	Agencies will need to determine the format for recording consanguinity.
013	Individual	Adoptive Status shall be recorded.	There was a general consensus that this concept should be represented for all individuals, given the implications adoptive status has for risk assessment.	Adoptive Status	Yes	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
014	Individual	Disorders shall be recorded.	The group felt that developing a specific list of disorders that should be queried about by all EHR/PHR systems was beyond the scope of this work. There was a feeling that prompts for disorders where family health history data has significant affect on risk assessment is of value, particularly if a given application is to be patient completed. However it was recognized that the list would need to be context specific and would be best determined by the end user based on their patient population. It was noted by members that a standardized vocabulary should be used for disorders recorded in the family health history section.	Disorders	Yes	Agencies will need to determine the format for documenting disorders, whether from a canned list, or free text, or a combination.
014.1	Individual	Disorder diagnosis shall be recorded.	Entry of multiple diagnoses for multiple disorders shall be allowed.	Disorder Diagnosis	Yes	Agencies will need to determine the format for documenting diagnosis (ICD/free text/canned list)
014.2	Individual	Diagnosis onset shall be recorded.	Age of disorder onset.	Onset	Yes	
014.3	Individual	Capability to document multiple disorders including separate occurrences of the same disorder for each individual.		Various	Yes	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
015	Individual	Capability to collect research identifier (placeholder).	A blank placeholder to be assigned a value only if individual is part of data bank/research protocol. This would likely be a number assigned by an entity interacting with the data base around which the EHR/PHR is structured.	Research Identifier	Yes	
016	Individual	Relevant laboratory data shall be recorded.	Refers to genetic test results relevant to family history, including mutation data about disorders in the family.	Lab Test Name/ Result/Result Date	Yes	Agencies will need to determine specific lab test results to be included and the format for collecting the data.
017	Individual	Relevant genetic test data shall be recorded.	There was a sentiment that there should be some capability to represent genetic laboratory data in the context of the family health history. It was felt that this information is critical for risk assessment.	Genetic Test Name/Result/ Result Date	Yes	Concern was raised about representing tests that were not strictly genetic (e.g., cholesterol) in the family health history section, especially because it might be difficult to provide adequate guidance on what tests results are truly genetic. Another concern voiced was the need for verification of the accuracy of this information if acquired directly from the patient and issues around confidentiality and privacy.

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
018	Individual	Capability to indicate "Unknown" for data entry fields.	In the case where the question is asked, but the answer is not known.	Unknown Indicator	Yes	Need to determine if this capability will be restricted to only certain data fields, and if so, which ones.
019	Individual	Capability to indicate approximate dates/ages for data fields.	This was felt to be a critical capability given that patients frequently have difficulty recalling specific dates/ages, and that this information would affect risk assessment if left blank. Most risk assessment utilizes age ranges or cut-offs, so the absence of a specific age will not significantly impact risk stratification.	Approximation Indicator	Yes	Need to determine if this capability will be restricted to only certain data fields, and if so, which ones.

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
020	Individual	Capability to designate fields or data as "Sensitive."	There was discussion regarding whether or not there should be a mechanism to designate sensitive fields (e.g., fields that the patient or provider would prefer to restrict access to). In general it was felt that this capability is very important, particularly when considered in the context of potential data sharing between individual EHR/PHR users. Further the group felt that the issue is broader than the topic of family health history and is being addressed by other entities. There was no clear consensus that this group should attempt to define the particular elements that a user may consider sensitive.	Sensitive Data Indicator	Yes	
021	Individual	Capability to capture certainty of data.	Consensus is that certainty of data determinations might be best left to the clinician to note, when appropriate, in a text box.	Certainty of Data (text)	Yes	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
022	Individual	Capability to share data between EHR systems and other points of data sharing, such as providing data to bio-banks in the context of the EHR, or between relatives in the PHR environment.	The general consensus was that means should be developed to provide the patient the maximum control over who the data is shared with and what data can be shared.	Various	Yes	Some group members discussed the possibility of linking relative's data at the level of the EHR which would permit automatic population of the patient's family health history directly from the medical record. The group recognized that attaining this level of integration would require significant changes in both law and cultural norms.
023	Individual	Capability to make annotations in a text box for various data entry fields.	Felt to be critical to have the ability to make notations for information that does not fall into structured data elements, but is felt to be critical to risk assessment.	Various	Yes	Agencies need to determine which data entry fields should allow text entry, either as a stand alone entry, or as a supplement to canned data entry.
024	Individual	Capability to capture place of birth.	Not thought to be helpful to current risk assessment strategies.	Place of Birth	No	
025	Individual	Capability to capture date of death.	Limited by patient recall	Year/Full Date	No	
026	Individual	Capability to capture multiple-birth status for relatives.	Not thought to be as valuable for relatives as for the patient.	Multiple-Birth Status	No	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
027	Individual	Capability to capture assigned gender.	There was a consensus that though gender issues are potentially relevant to disease risk assessment, that these are relatively rare cases and could be handled by the use of text box annotations.	Assigned Gender	No	
028	Individual	Capability to capture consanguinity for relatives other than parents.	Thought to be most relevant to record for the parents of the patient, rather than other relatives.	Consanguinity for Relatives	No	Agencies will need to determine the format for recording consanguinity.
029	Individual	Capability to capture disorder severity.	Not felt by the group to be easily quantified, nor terribly relevant to disease risk prediction.	Disorder Severity	No	
030	Individual	Capability to represent non-diagnosis health status.	The ability to represent the health status as “poor”, for example, absent other information was not thought to be helpful for risk assessment.	Health Status	No	
031	Individual	Capability to capture partner status of patient.	For patient, can be pulled from record, if existing, and felt not to be helpful to disease risk assessment if included for relatives.	Partner Status	No	
032	Individual	Capability to capture non-genetic laboratory data.	Numerous concerns were raised about representing lab results which were not clearly genetic in the context of family health history.	Lab Test Name/Result/ Result Date	No	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
033	Individual	Capability to capture relevant environmental data.	Thought to be potentially very important, however the consensus was that this information should be pulled for the patient from other parts of the health record, if existing. For relatives this approach is obviously problematic. Questions were raised about the validity of patient reported data regarding relevant exposures.	Environmental Data	No	
034	Individual	Capability to capture relevant social data.	For patient, can be pulled from record. The group felt that some social data might best be represented in the family health history section, such as shared households, quality of family relationships. The consensus was that the impact of this information on disease risk assessment was unclear and that this is a complex issue beyond the scope of this group's efforts.	Social Data	No	
035	Individual	Capability to tailor family health history data collection in a context-sensitive structure.	The group recognized the potential value of systems that could tailor questions asked about family health history to the demographics of the patient. However, it was felt that this was best left up to the creator of the EHR/PHR as the questions could potentially be very dependent on the population being targeted.	Various	No	

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
036	Family	Capability to record and represent free text or tabular data following structured data collection standards		Various	Yes	
037	Family	Capture data that allows for generation of a pedigree.	Even if the system cannot itself generate a pedigree, the data needed for manual generation should be available. If the system does generate a pedigree the representation should adhere to a standardized graphical nomenclature.	Various	Yes	Specific links must be defined so the appropriate data entry fields can be programmed to allow for generation of a pedigree.
038	Family	Capability to redefine the index case.	It was felt that this function – the ability to redefine the person about which the history/pedigree is constructed – is important for the PHR and could facilitate elective exchange of family health history information outside of the health care environment.	Various	Yes	
039	Family	Capability to record information on 1st and 2nd degree relatives including: • Mother/father • Siblings • Children • Aunts/uncles • Cousins • Grandchildren • Nieces/nephews	The group consensus was that the system should be capable of handling as many relatives as possible. It was felt that persons completing a family health history (clinicians or patients) should be encouraged to provide information on a minimum of 1st and 2nd degree relatives.	Family health history data for each relative.	Yes	Need to define what specific information should be recorded for each type of relative.

ID	Focus	Requirement	Clarification	Data Item(s)	Required	Follow-Up
040	Family	Capability to add free text data entry for each relative.	Use of text box to note special cases. Examples include gamete donor/surrogate, step parents, half siblings, etc.	Text	Yes	
041	Family	Capability to document relationship qualities for each relative including: • Estranged/difficult • Close (emotionally/ financially) • Household member	Though there was a general recognition that there is value in representing aspects of the family health history that are not strictly related to inheritance in the pedigree, the feeling was that this capability should be optional at this time.	• Estranged/ difficult • Close • Household member	No	There will need to be ongoing discussions about the best way to represent this information in the health record. Also discussed was the fact that this information could be pulled from the record if needed for clinical decision support. Text box annotation could be used where the patient or clinician felt the information to be particularly important.

7.0 Glossary

Biobank: A biobank, also known as a biorepository, is a place that collects, stores, processes and distributes biological materials and the data associated with those materials (From: http://www.biobankcentral.org/importance/what.php accessed October 2007).

Clinical Decision Support (CDS): Clinical decision support "…refers broadly to providing clinicians and/or patients with clinical knowledge and patient-related information, intelligently filtered, or presented at appropriate times, to enhance patient care." (Teich JM et al. Clinical Decision Support in Electronic Prescribing: Recommendations and an Action Plan Report of the Joint Clinical Decision Support Workgroup. J Am Med Inform Assoc. 2005 Jul–Aug; 12(4): 365–376.)

Consanguinity: Refers to the fact that two persons have a common, recent ancestor. For example, unions/marriage contracted between second cousins or closer are categorized as consanguineous.

Continuity of Care Record (CCR): CCR is a health record standard specification developed jointly by ASTM International, the Massachusetts Medical Society, the HIMSS , the American Academy of Family Physicians, the American Academy of Pediatrics, and other health informatics vendors. The CCR standard is a patient health summary standard. It is a way to create flexible documents that contain the most relevant and timely core health information about a patient, and to send these electronically from one care giver to another.

Electronic Health Record (EHR): The electronic health record is a longitudinal electronic record of patient health information generated in one or more encounters in any care delivery setting. This information may include patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, laboratory information and radiology reports (From: PHC Prototype Use Case).

Genomics: Genetic/genomic services are types of health services provided by laboratories and various health providers, including primary care physicians, medical geneticists, pathologists, genetic counselors, and genetic nurses. They include laboratory services that involve the provision of tests using genetic/genomic technologies, interpretation of results, and oversight of the test's performance. Other genetic/genomic services include identification or diagnosis of individuals and families at risk for a disorder with a genetic component or who could benefit from pharmacogenomic testing. They also include provision of support and genetic counseling to patients, facilitation of genetic/genomic testing, assistance with the interpretation of test results, explanation of germline, inherited and acquired disorders, analysis of inheritance patterns, review of the potential options for intervention, and management of clinical treatment (From: The draft SACGHS Coverage and Reimbursement Report).

Health Information Technology Standards Panel (HITSP): The Healthcare Information Technology Standards Panel is a private multi-stakeholder coordinating body designed to

provide the process within which affected parties can identify, select, and harmonize standards for communicating healthcare information throughout the healthcare spectrum. HITSP functions as a partnership of the public and private sectors and operates with a neutral and inclusive governance model administered by the American National Standards Institute (From: http://www.himss.org/ASP/topics_hitsp.asp).

Office of the National Coordinator (ONC): Office of the National Coordinator for Health Information Technology, formed in 2004 within the US Department of Health and Human Services. The mission of this office is to achieve widespread adoption of interoperable electronic health records in the US within 10 years (From: http://www.hhs.gov/healthit/news/).

Pedigree: A pedigree is a graphic, visual presentation of a family's health history and genetic relationships for the purpose of health risk assessment. It provides, at a glance, the distribution of a medical condition in a group of close relatives. If the condition clusters among relatives or follows a clear pattern of inheritance, then the risk for the condition can be assessed for the unaffected family members.

Personalized Health Care (PHC): Personalized health care describes medical practices that are targeted to individuals based on their specific genetic code in order to provide a tailored approach. These practices use preventive, diagnostic, and therapeutic interventions that are based on genetic tests and family health history information. The goal of personalized health care is to improve health outcomes and the health care delivery system, as well as the quality of life of patients everywhere (Accessed from: PHC website).

Personal Health Record (PHR): A health record that can be created, reviewed, annotated, and maintained by the patient or the caregiver for a patient. The personal health record may include any aspect(s) of the health condition, medications, medical problems, allergies, vaccination history, visit history, or communications with health care providers (From: PHC prototype use case).

Proband/Index Case: The affected individual through whom a family with a genetic disorder is ascertained; may or may not be the consultand (the individual presenting for genetic counseling) (From: http://ghr.nlm.nih.gov/ghr/glossary/proband).

Zygosity Testing: The process through which DNA sequences are compared to assess whether individuals born from a multiple gestation (twins, triplets, etc.) are monozygotic (identical) or dizygotic (fraternal); often used to identify a suitable donor for organ transplantation or to estimate disease susceptibility risk if one sibling is affected (From: Genetics Home Reference, National Library of Medicine http://ghr.nlm.nih.gov/ghr/glossary/zygositytesting).

8.0 Appendix A

July 31, 2007

The Honorable Michael O. Leavitt
Chairman
American Health Information Community
200 Independence Avenue, S.W.
Washington, D.C. 20201

Dear Mr. Chairman:

The American Health Information Community (AHIC) has given the following broad charge to the Personalized Health Care Workgroup:

> **Broad Charge for the Workgroup:** Make recommendations to the AHIC for a process to foster a broad, community-based approach to establish a common pathway based on common data standards to facilitate the incorporation of interoperable, clinically useful genetic/genomic information and analytical tools into electronic health records to support clinical decision-making for the clinician and consumer.

The Workgroup's deliberations have highlighted a number of key issues regarding the broad charge, including the following:

1. Genetic/Genomic Tests
2. Family Health History
3. Clinical Decision Support
4. Confidentiality, Privacy, and Security

This letter provides both context and recommendations for how the issues of genetic/genomic tests and family health history can be addressed in the next twelve months.

BACKGROUND

The Workgroup's vision of Personalized Health Care (PHC) is a consumer-centric system in which clinicians and consumers work together to customize diagnostic, treatment, and management plans based on a variety of factors, including the consumer's culture, personal behaviors, preferences, family health history, and the individual's unique genetic/genomic makeup. In this desirable future, consumers and clinicians both have ready access to information needed to identify and assess individualized treatment options as well as the resources and reimbursement mechanisms necessary to support implementation of a more extensive menu of tests and treatments.

Underpinning this vision is the confluence of two powerful forces, the development of Health Information Technology (HIT) and the rapid advances in the basic understanding of the

relationships between health, disease, genetics/genomics, and prevention and treatment options. Knowledge of an individual's genetic/genomic makeup appears to have an exceptionally powerful ability to assist with disease prediction, diagnostic accuracy, targeted treatments, medication dosing, and health management.

The PHC Workgroup has held six meetings since its formation in October 2006. Testimony from a wide variety of experts in standards development, genetics/genomics, laboratory testing procedures and systems, privacy concerns, tools and standards for family health history, and commercial and government electronic health record (EHR) systems has informed the Workgroup's discussions. In March 2007, the Workgroup developed a vision of PHC from four perspectives: the consumer; the clinician; the researcher; and the health plan/payer. Following this visioning session, the Workgroup outlined its priorities in the areas of: genetic/genomic tests; family health history; clinical decision support; and confidentiality, privacy, and security. The vision summary and priorities documents were presented to the AHIC on April 24, 2007. Subgroups of the Workgroup were formed to address each of these four priority areas. Two of these subgroups, genetic/genomic tests and family health history, have developed recommendations that are being advanced to the AHIC by the PHC Workgroup.

If accepted by the AHIC, these recommendations should be considered for adoption by the Department of Health and Human Services (HHS) as HHS policy regarding current and future federal activities as they relate to the Workgroup's charge.

INITIAL RECOMMENDATIONS

I. Overarching

With the completed sequence of the human genome, genetic/genomic testing and its possibilities have moved from the sidelines into mainstream medicine. There are over 1,400 diseases for which genetic/genomic tests are used in current clinical practice, and several hundreds more are available in a research setting.[i] A genetic/genomic test can be performed on a wide variety of tissue samples and across the human lifespan, providing information on predispositions for a disease, presence of a disease, the risk of passing a disease onto offspring, and potential positive or adverse responses to therapeutic interventions.

In addition to the increasing adoption of genetic/genomic testing in medical practice, clinicians have always used a basic and important genetic/genomic tool in everyday practice: family health history. Combined with the power of genetic/genomic testing results, family health history adds value and provides useful predictive information. Broadly stated, genetic/genomic information has the potential to identify and predict the health outcomes of individuals and their families.

[i] http://www.genetests.org/

Consumers today are concerned that their health information may be used for unintended purposes or without their authorization. Compounding this concern are the limited understanding of new genetic/genomic tests for heritable disorders, the immutability of this information across the consumer's entire lifetime, the predictive abilities attributed to genetic/genomic information, and the potential for unintended informing of relatives because of a common genetic/genomic background. However, if consumers avoid genetic/genomic tests because of fear, they are potentially at risk by not having information available to them that could substantially and beneficially alter their health care. Therefore, maintaining the public's trust in the use of their personal health and genetic/genomic information, by developing technical and policy guidelines to ensure the security of their genetic/genomic data, is key to maximizing utility and health benefits. Consumer authorization of access to their genetic/genomic information should be taken into consideration as these use cases are developed. Therefore, the PHC Workgroup will work with the Confidentiality, Privacy, and Security (CPS) Workgroup to consider if aspects of genetic/genomic test results and family health history information may raise special concerns about confidentiality, privacy, and security relative to other types of medical data.

The Workgroup identified the following actionable recommendations for the next twelve months that begin to address one aspect of the broad charge, incorporating clinically useful genetic/genomic information into the EHR.

> **Recommendation 1.0**: The Community should advance the area of Personalized Health Care as a Priority for Use Case Development.
>
> **Recommendation 1.1**: Priorities for use cases in the area of Personalized Health Care should be developed in conjunction with work performed by the genetic/genomic test workgroup and the family health history workgroup described in Recommendations 2 and 3. The use cases should additionally leverage the work in related activities including: the AHIC EHR, CPS, and Consumer Empowerment (CE) Workgroups; the Harmonized Use Case for Electronic Health Records (Laboratory Results Reporting); the Consumer Access to Clinical Information Use Case; and others.

II. Genetic/Genomic Tests

Inclusion of genetic/genomic test results in the EHR or personal health record (PHR) could enable the personalization of health care decisions through avoidance of adverse reactions, selection of optimal interventions, and beginning the transition of the health care sector from a reactive to a predictive enterprise. Standardized electronic recording of data associated with laboratory performance of genetic/genomic tests and, in parallel, inclusion of relevant results from genetic/genomic tests in the EHR have been identified as immediate priorities for recommendation by the PHC Workgroup.

Genetic/genomic testing in humans generally falls into two categories: molecular and biochemical. A molecular genetic/genomic or cytogenetic test may be defined as an analysis performed on human DNA, RNA, and chromosomes to detect heritable or acquired disease-related genotypes, mutations, or karyotypes for clinical purposes. A biochemical genetic/genomic test may be defined as the analysis of human proteins and certain metabolites, which are predominantly used to detect inborn errors of metabolism, heritable genotypes, or mutations for clinical purposes. Tests that are used primarily for other purposes, but may contribute to diagnosing a genetic/genomic disease (e.g., blood smear, certain serum chemistries), would not be covered by this definition.[ii]

The process of performing a genetic/genomic test can be segmented into three distinct phases with each having different information collection requirements. The three phases include: (1) the pre-analytic phase, which encompasses such events as determining which genetic/genomic test, if any, is appropriate to answer the clinical question being asked, collecting clinical information that is necessary to interpret the test, and collecting an appropriate sample and transporting it to the test site; (2) the analytic phase, which involves steps taken to perform the analysis and analyze the results; and (3) the post-analytic phase, which includes reporting and interpretation of the results.[ii]

As the area of genetic/genomic tests is relatively new to the medical community, and there are a growing number of different types of tests that are captured by the broad definition of a genetic/genomic test, standards development in some areas of this diverse category may be immature. Therefore, an iterative process should be pursued where use case development is performed in parallel with standards identification/creation. Gaps in available standard reference materials, protocols, metrics, IT standards (terminology, coding, messaging, instrument integration, and implementation guides) will therefore be highlighted early in the process and brought to the attention of the appropriate standards development organizations. Standards that address communication between EHRs and Laboratory Information Systems (LIS) are crucial to ensure comprehensive bidirectional transfer of information between the EHR and LIS in the pre- and post-analytic phases.

The many different information requirements for incorporation of genetic/genomic test information in the EHR is an issue of immediate concern to the PHC Workgroup. Longer term goals of this Workgroup include supporting the development of accompanying information about benefits, risks, analytical validity, clinical validity, and clinical utility to ensure the development of robust clinical decision support concerning genetic/genomic test results. Additionally, incentives to develop new genetic/genomic tests that provide new or added value to clinical care and the corresponding reimbursement strategies to ensure their widespread use need to be addressed. These longer term goals would be facilitated by the development of means and standard materials and processes for capturing laboratory data and test results identified as the immediate concerns for Healthcare Information Technology Standards Panel (HITSP) use case

[ii] CDC definition, Federal Register, Vol 65, No 87, 5/4/2000, 25928.

development. Future recommendations by the PHC Workgroup may address these longer term issues.

> **Recommendation 2.0**: An extension to the Harmonized Use Case for EHRs (Laboratory Results Reporting) should be developed to address the specific information needs in the pre-analytic, analytic, and post-analytic phases of genetic/genomic tests. This extension to the use case should additionally address the need for integrated data flow across the pre-analytic, analytic, and post-analytic phases of genetic/genomic testing and address both the EHR and Laboratory Information Systems.
>
> **Recommendation 2.1**: A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to identify what types of data and information are generated when performing genetic/genomic tests, and to identify standard metrics, terminology, language, and processes. This work should inform the extension to the Harmonized Use Case for EHRs (Laboratory Results Reporting) developed for genetic/genomic tests.
>
> **Recommendation 2.2:** Research activities that increase the knowledge base regarding genetic/genomic test results need to be supported. The National Institutes of Health (NIH) should continue to work with public and private partners to support, develop, and enhance public reference databases that enable more effective and efficient genetic/genomic testing and incorporation of test results that can be aggregated in electronic health records.[iii]

III. Family Health History

Health care professionals and the general public have widely accepted the importance of family health history for predicting increased risk for a number of common diseases, including cancer, heart disease, and diabetes. As our scientific understanding of the molecular and genetic/genomic basis for health and disease improves, the importance of family health history as a valuable predictive tool has only increased. This has been highlighted throughout HHS by the Surgeon General's online web portal for collecting family health history information, the 'My Family Health Portrait', developed in conjunction with the NIH and the Centers for Disease

[iii] Specifically, NIH, and the National Library of Medicine (NLM) in particular, should continue to: (1) enhance its collection of mutation data; (2) expand a National Center for Biotechnology Information (NCBI) clinical reference sequence database (RefSeqGene); (3) expand coverage of genetic/genomic tests in Logical Observations Identifiers Names Codes (LOINC) in collaboration with other HHS agencies, state public health laboratories, and the American Society of Human Genetics; and (4) provide more integrated access to genetic/genomic information for the public through NCBI portal developments, the Genetics Home Reference, Online Mendelian Inheritance in Man (OMIM), and MedlinePlus in cooperation with other HHS agencies, the Genetic Alliance, the American College of Medical Genetics, and other professional and disease advocacy groups.

Control and Prevention. The Family Health History priority area for the PHC Workgroup includes activities of immediate concern related to use case development by HITSP. The use case should represent the continuum of information collection, from consumer entry of family health history in the PHR to clinician entry of family health history in the EHR, with the longer term goal of interoperability between the PHR and EHR. Health care providers involved in any pilots of this use case should examine the merits of developing a modular family history tool, where collection of family health history is performed within the EHR, followed by messaging of this information to a variety of richer family history tools that perform risk analyses. In these tools, family history data can continue to be extended with new family history information as well as analyzed using the latest risk assessment algorithms. The enhanced family history and results of these algorithmic calculations could then be returned to the EHR, allowing for the ongoing curation of novel risk assessment algorithms and use of these tools in concert with well established family health history collection tools.

Additionally, the longer term goals of the Family Health History priority include: infrastructure and incentives to use PHRs to improve consumer-clinician communication; and characterization of the validity and utility of use of family health history in making clinical decisions. An overarching theme across the Family Health History priority area is how the clinician can use the family health history information, and this should be considered in short and long term activities. These longer term goals are contingent on the development of means and standards to capture the family health history information identified as the immediate concerns for HITSP use case development. Future recommendations by the PHC Workgroup may address these longer term issues.

> **Recommendation 3.0**: A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to develop a core minimum data set and common data definition available for primary care collection of family health history information.
>
> **Recommendation 3.1:** Additionally, studies should be performed as part of this collaboration as an evidence-base to determine the validity and utility of family health history risk assessment and management tools, clinical decision support tools, and how clinicians view this information as helpful for informing their medical decisions.
>
> **Recommendation 3.2**: Federal agencies in conjunction with private health care organizations with similar interests and expertise sponsoring pilots in the area of family health history should be used to evaluate the core minimum data set and evidence-base developed through Recommendations 3.0 and 3.1. Health care providers involved in these pilots should also examine the feasibility of consumer-clinician exchange of family health history information between PHR and EHR systems. When possible, the pilots should test and implement the standards and architecture identified in the HITSP developed use case.

These recommendations are supported by information obtained through research and testimony to the Personalized Health Care Workgroup, which is contained in the supporting documents available at http://www.hhs.gov/healthit/ahic.

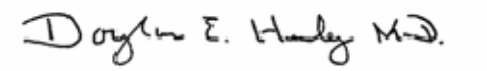
Thank you for giving us the opportunity to submit these recommendations. We look forward to discussing these recommendations with you and the members of the American Health Information Community.

Sincerely yours,

John Glaser

Co-chair, PHC Workgroup

Sincerely yours,



Douglas E. Henley

Co-chair, PHC Workgroup